MED13L gene

mediator complex subunit 13 like

Normal Function

The *MED13L* gene provides instructions for making a protein that is one piece (subunit) of a group of proteins known as the mediator complex. This complex regulates the activity (transcription) of genes. Transcription is the first step in the process by which information stored in a gene's DNA is used to build proteins.

The mediator complex physically links the proteins that can turn genes on, called transcription factors, with the enzyme that carries out transcription, called RNA polymerase II. Once transcription factors are attached to RNA polymerase II, transcription begins.

Researchers believe that as part of the mediator complex, the MED13L protein is involved in many aspects of early development, including development of the heart, nerve cells (neurons) in the brain, and structures in the face. The mediator complex plays a role in several chemical signaling pathways within cells. These pathways help direct a broad range of cellular activities, such as cell growth, cell movement (migration), and the process by which cells mature to carry out specific functions (differentiation).

Health Conditions Related to Genetic Changes

MED13L syndrome

More than 50 mutations in the *MED13L* gene have been found to cause *MED13L* syndrome. This condition is characterized by moderate to severe developmental delay and intellectual disability and minor differences in facial features. Additionally, some people with *MED13L* syndrome have recurrent seizures (epilepsy) or heart abnormalities that are present from birth (congenital heart defects).

Some *MED13L* gene mutations insert or delete regions of DNA within the *MED13L* gene. These genetic changes lead to a reduction in the total amount of MED13L protein in cells. Other mutations change single protein building blocks (amino acids) in the MED13L protein. It is thought that the altered protein interferes with the function of the normal protein produced from the non-mutated copy of the *MED13L* gene (such mutations are described as "dominant-negative"). Because dominant negative mutations impair the function of proteins made from both the altered copy of the *MED13L* gene and the normal copy, individuals with dominant negative mutations tend to have more severe signs than people with mutations that affect protein production from just the altered copy of the gene. While it is likely that mutations in the *MED13L* gene impair the control of gene activity by the mediator complex, it is

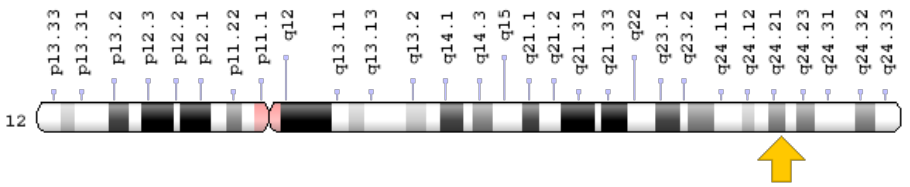
unclear how these changes lead to the particular cognitive and physical features of the disorder.

Critical congenital heart disease

Chromosomal Location

Cytogenetic Location: 12q24.21, which is the long (q) arm of chromosome 12 at position 24.21

Molecular Location: base pairs 115,958,576 to 116,277,693 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA1025
- MEDIATOR COMPLEX SUBUNIT 13-LIKE
- PROSIT240
- PROTEIN SIMILAR TO TRAP240
- THRAP2
- THYROID HORMONE RECEPTOR-ASSOCIATED PROTEIN 2
- TRAP240-LIKE PROTEIN
- TRAP240L

Additional Information & Resources

Educational Resources

- Autoimmunity: From Bench to Bedside: Transcription https://www.ncbi.nlm.nih.gov/books/NBK459456/#gene.s4
- Centers for Disease Control and Prevention: Facts about dextro-Transposition of the Great Arteries (d-TGA) https://www.cdc.gov/ncbddd/heartdefects/d-tga.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MED13L%5BTIAB%5D%29+OR+%28mediator+complex+subunit+13+like%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- MEDIATOR COMPLEX SUBUNIT 13-LIKE
  http://omim.org/entry/608771
- TRANSPOSITION OF THE GREAT ARTERIES, DEXTRO-LOOPED 1
  http://omim.org/entry/608808

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MED13L.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MED13L%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:22962
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:23389
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/23389
- UniProt
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